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# Living With and Treating Rare Diseases: Experiences of Patients and Professional Health Care Providers

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## Abstract

We explored the experiences of illness of patients suffering from rare diseases and of the health professionals who care for them at the Center for the Interregional Coordination of Rare Diseases of Piedmont and Valle d'Aosta in Italy. The research was carried out between 2010 and 2011. We collected qualitative data from 22 patients and 12 health professional health care providers. The interviews were analyzed using the Colaizzi phenomenological approach. We identified five themes from the narratives of the patient participants—dealing with disease development, living with the disease, everyday living, relating to others, and relations with health care providers—and four themes from the professional health care participants—dealing with the disease, dealing with expectations, building relationships, and being operators in the context. The study has raised awareness about the issue of rare diseases and it provides some useful considerations for improving services.

**Keywords** health care, long term; lived experience; phenomenology; illness and disease; qualitative analysis

Rare diseases cover a heterogeneous set of pathologies that can strike any organ or system of the human body. In literature, there are different definitions: the European Union defines as a rare disease a pathology that affects five or fewer individuals out of 10,000 in the European population; instead, in the United States a rare disease is one that affects less than 200,000 individuals in all the nation (Wästfelt, Fadeel, & Henter, 2006).

From the existing estimates, the number of disorders in the category of rare disease is very high (> 5,000 according to the WHO). These are diseases that can strike between 6% and 8% of the population in their lives. Thus, even though the prevalence of these pathologies is low, the total number of people affected is about 30 million in the European Union and about 25 million in the United States (Agazio et al., 2005; Schieppati, Henter, Daina, & Aperia, 2008). Most of these disorders have a genetic origin, while the rare diseases whose origin is not genetic can instead be the result of a bacterial or viral infection, or it may depend on environmental causes, allergy, or have a proliferative basis (Faurisson & Kole, 2009). Despite the high number of pathologies covered by the definition, it is possible to identify some common features: They are diseases of a chronic, degenerative nature; they may involve severe disability and heavily compromise the quality of life. Such diseases require continuous specialist assistance. Patients with rare diseases have common experiences as regards delays in diagnosis, difficulties in access to treatment and care, and the loss of economic and social opportunities. Because of the very small numbers of cases and their dispersion, scientific research has shown little interest in these pathologies owing to the difficulties involved in organizing clinical studies for identifying their causes and studying their development, and in amortizing the costs of a specific pharmacological study. Europe has tried to protect the rights of these patients by issuing regulations and recommendations. The Member States have been invited to draw up and put into effect appropriate strategies and measures to guarantee patients access to high-quality health care, including diagnostic tools and treatment, and to promote and foster research in this field. Moreover, the European Union has promoted in each nation the identification of Centers of competence for rare diseases that participate in the European networks of reference (European Parliament and the Council of the European Union, 2000; Council of the European Union, 2009). In Italy, the national network of rare

diseases was established by Decree n. 279/2001 (Italian Ministry of Health, 2001). This is a clinical and epidemiological network composed of accredited Centers for the prevention, surveillance, diagnosis, and treatment of rare diseases that are specifically identified by the regional health authorities. The aims of the network are to promote the spreading of information and documentation on rare diseases, facilitate access to specialist care for patients, and provide consultancy and support to the doctors of the Italian Health Service (Decree n.279/2001).

People who may have a rare disease go to these Centers to find a diagnosis and appropriate treatment. As these pathologies are by nature chronic, potentially invalidating, with a clinical course that alternates between periods of acute flares and phases when the disease is in remission, patients are followed for a very long time, also for the duration of their lives, coming into contact with various types of professional health care providers who accompany them in their treatment program. The medical treatment currently available for these patients is unlikely to clear up the disease. These conditions can create an experience of suffering in patients undergoing treatment, heightening the feelings of isolation that they already have on account of the rare nature of their condition, and they can give rise to a profound sense of impotence in the professional health care providers. There is therefore the need to build a meaningful relationship between the person with the rare disease and the health care staff.

All these considerations led us to set up a research project in one of the Regional Centers of Reference for Rare Diseases. To raise awareness of the experiences of patients and professional health care providers can be a useful strategy for identifying elements for improving the quality of the health services provided (Knight & Senior, 2006).

The theoretical framework of the project is provided by narrative medicine (Charon, 2001, 2004, 2006; Greenhalgh & Hurwitz, 1998). Acting as a bridge between the clinical knowledge of the physician and that of other professional health care providers and the patients' subjective experiences, narrative medicine aims to "accompany" the various stakeholders toward the shared management of the disease. This approach assumes that the narration constitutes the ordering element of the experience and allows the structuring of the continuity of subjective experience.

We realized our study using narrative medicine as a tool for improving the quality of care through the analysis of the narratives of the patient participants and professional health care provider participants, restructuring them in a shared interpretative framework. The aim of this study has been both to explore the impact of the rare disease on the patients' lives and together with the experience of the health professionals who look after these patients on a daily basis, and also to highlight the positive and the critical aspects of the health care services.

## **Method**

### **Participants**

We carried out the study in a Regional Center of Reference for Rare Diseases (CMID; Multidisciplinary Center of Immunopathology and Documentation of Rare Diseases). It supports a model based on the promotion of generally agreed diagnostic and treatment health care protocols, on the swift diffusion of information on health and legislation and on the use of the Interregional Register of Rare Diseases as a tool for identifying problems such as delayed diagnosis, inappropriate treatment approaches, and the areas where it is necessary to invest greater resources. The Center is composed of an outpatients department

where new patients are examined and regular patients have their routine check-up, and a day hospital for those who need infusion therapy or invasive treatment as they are in an acute phase of the disease.

The activity of the Center is based on multiple interdisciplinary cooperation among specialists (immunologists, clinical pathologists, hematologists, nephrologists, rheumatologists, infectious disease specialists, geneticists) and various professional health care providers (doctors, nurses, physiotherapists, laboratory technicians, health and social workers).

We have chosen purposive sampling by asking a group of patients and professional health care providers at the Center to participate. In the study, there are patients with rare diseases according to the European Union definition of the year 2000: diseases with prevalence equal or inferior to 5 cases per 10,000 people in the EU. Patients participating in the study have to have been registered for at least one session in the day hospital and are still undergoing active follow-up, whereas the professional health care provider participants (doctors, nurses, health and social workers) must have worked at the Center for at least 6 months. The number of patient participants involved was established on the basis of reaching data saturation (Speziale & Carpenter, 2011). All of the professional health care providers at the Center were invited to participate. The Directorate-General of the Local Health Authority, to which the Center refers, and the Directorate of the Hospital Unit and the Director of the Department of the Center were all informed about the type of study, and consent was granted. The Institutional Review Board approved the study design and methods. Participation in the study was voluntary. Those contacted were informed about the aims of the study and the means of data collection. We pointed out the need to record the interviews. Participants were guaranteed anonymity and gave their written consent.

We interviewed 22 patients with different diagnoses of rare diseases (such as Scleroderma, Horton's disease, Wegener's granulomatosis, Mixed connective tissue disease, Addison's disease, Behcet's disease, Gaucher's disease) and 12 professional health care providers including health and social workers, nurses, doctors, and junior specialist physicians (see Table 1).

## **Procedures**

Narrative inquiry, which is a form of qualitative research that uses the collection of stories as its source of data, was adopted (Duffy, 2007). We used autobiographical narrative interviews to learn about the patient participants' and professional health care provider participants' experiences, according to the narrative inquiry approach. In this way, it was possible to show the voice of each participant and provide the opportunity to understand the complexity and depth of the phenomenon studied, revealing the interviewees' experiences, perceptions, and thoughts in their own words (Dicicco-Bloom & Crabtree, 2006; Giarelli, 2005).

The research team drew up two interview outlines, one for the patient participants and the other for the professional health care provider participants, with cues able to stimulate conversation and considerations with the interviewees, helping them to reconstruct their experience of the disease or of dealing with it:

The experience of living with a rare disease:

experience of first symptoms,

access to the health care and social services: asking for help,

experience of the time up to diagnosis (undergoing diagnostic tests, waiting, errors in diagnosis),

getting a diagnosis: impact on personal life,

finding the Rare Diseases Center,

receiving treatment/support (over time . . . , drugs, psychological support),  
living with a rare disease (relations with others, everyday life, work, changes),  
being a man/being a woman,  
the presence of associations.

The experience of caring for people living with a rare disease:

describe a rare disease,

the “patient” when they present at the center (the first meeting, expectations),

relating to patients seeking a diagnosis,

identifying the disease,

the quest for a diagnosis (reporting it to the person),

accompanying the treatment (offering support, being a point of reference, relating to the chronic aspect of the disease, dependence-independence, clinical deterioration, drugs),

the relationship with the patient and with his/her family/caregiver (people involved with the patient, understanding each other, difficult aspects/facilitative aspects).

The outlines were drawn up, starting with some models of disease history presented in the works by Giarelli (2005) and Wilcock, Brown, Bateson, Carver, and Machin (2003), and they were adapted to the context of the study.

The interview outlines were then reviewed by a group of professional health care providers at the Regional Center for Rare Diseases and by some patients. They were used as guides for the interviews. A series of open questions were also provided which were based on the interview outlines. These questions could be used if it was necessary to encourage the interviewee to reflect on or share his or her thoughts.

The interviews took place in a quiet, isolated room in the sole presence of the interviewer and the participant, and they were recorded. In one case, the interview with the patient took place in the presence of a family member. All of the interviews were conducted in Italian.

Between May and September 2010, there was a pilot study with four interviews with patients and four with professional health care providers. This initial phase of work made it possible to fine-tune the outlines for the interviews, identifying some more specific open questions to stimulate considerations on some aspects that emerged as being significant.

## **Data Analysis**

We adopted a phenomenological approach for this study. The aim of this research method is to describe the significance of the experience from the perspective of the individuals who have lived it to obtain greater knowledge of the nature and significance of that experience. The experience of the interviewees is the focal point of the phenomenological investigation as it is an element that is able to attribute meaning to the individual's perceptions of a particular phenomenon (Speziale & Carpenter, 2011).

The concepts of phenomenology essence, intuition, and reduction are the basis of the research method chosen. The essences are elements related to the ideal or real meaning of the phenomenon under study, in other words of those concepts that are able to identify a common knowledge of it (Fain, 2004; Speziale & Carpenter, 2011). Intuition represents the capacity of the researcher engaged in the description of the experiences to acquire an accurate and complete interpretation of it (Fain, 2004). Phenomenological reduction is a method of analysis that is able to guarantee an approach that is as free as possible from conceptual premises and the opportunity to describe the phenomena experienced as faithfully as possible. The reduction process fundamentally requires identifying any previous knowledge, idea, opinion, or belief

regarding the phenomenon being studied. Once all this has been identified it is necessary to separate it from the object of the investigation, in other words to “bracket” it. This allows the researcher to have an impartial approach and makes it possible to collect accurately what emerges from the data under examination (Speziale & Carpenter, 2011).

We analyzed the interviews using Colaizzi’s (1978) procedural interpretation as described by Polit and Beck (2013). Three different researchers analyzed the data separately and at the same time.

They carefully and repeatedly read all of the interviews to acquire a feeling for them. Then they reviewed each interview and extracted significant statements. They spelt out the meaning of each significant statement and organized the formulated meanings into clusters of themes, and after that they referred these clusters back to the original protocols to validate them. At this point, the three researchers compared their analyses to highlight and discuss any discrepancies and to reach an agreement. Then they integrated the results to formulate an exhaustive description of the phenomenon under study.

The quality of the study was ensured using the Lincoln and Guba framework (credibility, dependability, confirmability, transferability, and authenticity) as described by Polit and Beck (2013). Credibility and dependability were ensured through the use of an audit trail, verbatim transcription, and member checking. Where possible, the descriptions were returned to the participants for their confirmation.

## **Results**

We conducted the interviews between May 2010 and January 2011. Each interview, which lasted between 30 and 60 minutes, was recorded. The interviews with the patient participants and with the professional health care provider participants were analyzed separately. This was a deliberate choice so as to maintain the wealth of considerations that emerged, and as a result, the themes that arose were slightly different in the two groups.

The analysis of the texts of the interviews led to the identification of nine themes (five for the patient participants and four for the professional health care provider participants) with essential elements (see Tables 2 and 3). We chose some excerpts from the interviews to illustrate each theme. They are useful for understanding the participants’ experience, but they are not exhaustive of the many nuances that emerged. To guarantee the anonymity of the participants, the phrases are reported anonymously, accompanied by a code.

### **Results of the Interviews With the Patients**

#### **Dealing With Disease Development**

##### **Getting a diagnosis**

Various patients remember how difficult it was to get a diagnosis of their disease. Often the symptoms they felt were aspecific and therefore hard to place in the context of standard diagnosis procedures. This meant that they had to see several specialists and undergo various diagnostic tests:

I used to go to the Molinette hospital but we rather neglected (the disease), then the thing continued . . . I did not think it was a disease . . . I mean serious. Then my daughter met a woman on the train and talking she told her about the G . . . and a woman doctor examined me . . . then I went to the Molinette for a capillaroscopy. Then they told us that here there is the Rare Diseases Centre and I came here, and that was it! It took rather a long time before they discovered the disease . . . (Patient Participant 3)

For several patients, getting a diagnosis was experienced as a difficult process above all because they did not understand what was going on:

Quite frankly I did not know what it could be . . . I was a bit, in limbo. If at first I experienced it as “They are trying to find out,” when they admitted me to hospital in nephrology I had the feeling “If I’m here I’m really ill!” (Patient Participant 10)

### **Treatment approach**

With regard to the treatment, what emerges as fundamental—but not simple—is identifying the most effective drugs and finding the right balance in the dosage, taking into account the variability in the response of each individual patient.

Patients often refer to the side effects which are difficult to manage and which have a high impact on everyday life. In addition, they stress the obligation of having to periodically undergo treatment in hospital:

[In the period when the patient was taking 500 mg/die of cortisone] “I couldn’t sleep, I wasn’t sleepy, I was always frenetic. I couldn’t stay seated, I couldn’t even watch television. Sometimes I said to myself: ‘I don’t recognize myself!’ I irritated the others and it irritated me that the others would remain seated and still.” (Patient Participant 7)

It’s twice a month, not so often; but it’s . . . I don’t like it. It’s . . . you know, it’s the idea, isn’t it? Also because it creates dependence, in the end, and you can’t escape it. And . . . let’s say, you feel that a little bit of freedom has been taken away from you. (Patient Participant 4)

### **See progression between remission and relapse**

What characterizes the course of most of these pathologies is alternation between moments of stability when the symptoms of the disease are not evident and phases of relapse when the pathology reappears in an acute form and the patients feel the progression of the disease which they react to with anxiety and a sense of uncertainty about its possible evolution.

During the phases of relative well-being, the disease is not evident, it becomes a disease that “you can’t see.”

I think the problem is that quite rightly . . . for example, if a person has a broken arm you can see it! But we . . . our diseases—I speak for myself, but I have seen this with others too—no-one would think you had anything wrong with you. (Patient Participant 13)

### **Predicting the future**

This aspect crops up in many of the interviews. There are three elements: uncertainty about the more or less rapid or aggressive evolution of the disease, the fear of its worsening, and the hope that the stability obtained can be preserved.

Going forward . . . I am afraid of later, a little. I say to myself: “Perhaps now I am managing more. I am more . . . But later I say: I wonder how much longer it can hold out.” (Patient Participant 6)



## **Living With the Disease**

### **Adaptation strategies**

Patients report that they adopt different adaptation strategies for coping with the disease. They consider it effective to try and have a positive or optimistic approach to the disease and the treatment, trying as far as possible to lead a “normal” life:

The disease is part of us. Unfortunately we have to do all we can to resolve it with the support of the doctors, but it is basic . . . our reaction, our wanting to get better, never letting go. Thinking that there is always a way out. It may also be chronic, but it is a chronic that you can easily live with, because it is a very bearable chronic . . . once you can use your hands, be grateful. (Patient Participant 18)

Being able to accept one’s condition of disease, learning to live with it, turns out to be the most appropriate way to improve one’s quality of life, even though sometimes it may be necessary to change some habits.

What I am allowed to do. Every morning I get up really early to get an hour of time to do training: doing some everyday maintenance exercises, even though . . . my hands have deteriorated a lot. Above all I realize because in some exercises I cannot get up to the same speed that I used to. But it is also true that you can make up for it with the experience of expertise acquired over the years. (Patient Participant 21)

Patients often describe the work they had to do with themselves to activate the resources that are essential for dealing with the condition of disease. To find the inner strength to go on, to get up after each fall, and sometimes to turn a condition of weakness into one of strength:

At the beginning I was upset: I spent the days crying. Then in the end I looked at myself a moment and I said: “Yesterday I cried; and I did so the day before yesterday too, and also the other evening, and last month . . . the disease won’t go away. Indeed quite the contrary, I will just go and get worse at the cerebral level!” (Patient Participant 20).

### **The quest for independence**

Many of those interviewed stress the need to maintain areas of autonomy in their work and in their everyday activities, even when the limitations imposed by the disease make this difficult.

What often emerges is the patients’ difficulty in asking for and/or accepting help from the members of their family and friends or acquaintances. Patients experience this aspect as a potential loss of their independence and therefore as a defeat:

I have not been to the market by myself for two years. I always go in the company of my husband. And that brings on a nervous breakdown because I was used to doing it alone. I am not . . . I shall have to get used to asking. (Patient Participant 16).

### **The need to know the disease**

Often patients express their need to acquire information about their own disease independently. The most common means is looking up information on the Internet, but patients also use texts or take part in specific conferences on the subject:

I did my research by myself on internet, I downloaded all kinds of things, I have got that much stuff; and then the psychologist told me: “Now that’s enough! Indeed you already know too much about your disease!” (Patient Participant 18).

## **Everyday Living**

### **Functional limitation**

The limitations deriving from the condition of disease touch various aspects of everyday life: being able to move about autonomously and carrying out various activities of everyday life such as eating, sleeping, or managing holidays, free time, and recreation activities independently. Sometimes what is compromised is the chance to have an adequate social life.

But as it is now I’m taking it badly, because I can’t do anything, I get palpitations, I . . . I have my grandchildren and I can’t hold them in my arms . . . And nobody says anything to you! (Patient Participant 17)

### **Experiencing pain**

The experience of pain is widespread among those interviewed while the ways pain comes on, it’s perceived intensity and reactions to it are subjective. Often the experience is all-absorbing, concentrating patients’ attention and limiting everyday activity:

I can’t take it any longer: I did not even recognize my husband I was in so much pain; it was really unbelievable! (Patient Participant 19)

### **Influence on work**

For some of those interviewed as the disease progressed, it became incompatible with maintaining a job. For others, the condition was limiting for the development of their career or it was the reason for unwanted job reallocation to not very gratifying positions.

In practice this esophagitis, as I had to lift boxes, I used to bend down, which was very bad news! It was really something . . . And after a few years there was redundancy, and so my husband said: “Listen, as there is this redundancy, volunteer for it and you can stay at home.” I didn’t want to, I did not . . . And all in all, in the end I agreed to. (Patient Participant 2).

## **Relating to Others**

### **The role of the family**

The role of support played by the family emerges frequently in patients’ narrations. The role of support by the partner, children, and other family members is fundamental in helping the person with the disease to accept their condition and tackle daily difficulties more serenely.

Some patients express the fear that their need to be helped can be a burden for their families; others point out the changes that their state of illness has brought about in their relations with family members:

Sometimes I don’t say everything because I don’t want to burden them further; but my daughter, apart from the fact that she is a psychologist, is in a class apart as regards her understanding at a glance! In other

words she comes in, takes one look at me says: “Mum, you were in pain last night! You are walking a bit crooked . . .” Yeah, she observes me! (Patient Participant 18)

And then a thing that I miss is that . . . since I separated—and it is linked to my illness, because . . . with all my problems I distanced myself a lot from my ex-husband . . . I would have liked to have a child, but when you find yourself in certain situations . . . It’s a big decision to make! (Patient Participant 14)

### **Comparison with other patients**

The comparison with people with the same pathology is an element which crops up time and again in the narrations of those we interviewed. The patients observed themselves and sought their symptoms in other people. The comparison is sometimes deliberately sought, and at times it is avoided, as there is the fear of seeing possible negative developments. Often the patients in the waiting room will exchange their stories and experiences. This relationship may continue outside the treatment context (outside the surgery, clinic, or hospital). Patients often feel the need to contact associations that deal with patients with rare diseases.

One day there was a woman, here: first we were on the drip at the same time, I didn’t know what she had; I saw that she couldn’t open a bottle. It just slipped out when I said: ‘You can’t open it, do you want me to do it?’ Then the nurse came along and she opened the bottle. Then she asked me what was wrong with me, and I dared to ask her: ‘What have you got? Why can’t you open it?’ And she told me that she had scleroderma, she must have been about 40 years old . . . This woman talked . . . she talked! Then we also met up outside the hospital. (Patient Participant 16)

### **Relations With Professional Health Care Providers**

#### **Admission and follow-up**

Most patients express considerable satisfaction about how they are taken care of by the staff at the health care Center, and in particular what emerges is the positive memory of the welcome they received during their first meeting. Some stress the positive aspect of finding the answer to their multiple problems in the team present in a single center.

Then he said: “Listen, now if you agree I would admit you to be followed by our team!” Now I nearly burst into tears I was so happy! Because I said: “Look, I could ask for nothing more as I really don’t know what on earth to do at this point!” (Patient Participant 15)

Some patients point out some critical points with regard to their admission. They have a negative view of the turnover of the doctors who are still specializing, as it makes them feel that there is discontinuity in their admission and follow-up.

My only . . . criticism is that it doesn’t make sense for me to see . . . all the doctors leave. When they get to know you, they know about your problems and then you have to start all over again. Perhaps you have built up a certain degree of familiarity with the doctor and then you get another one. (Patient Participant 10)

#### **Features of the relationship**

In their interviews, the patient participants often refer to the skill shown by professional health care providers in establishing positive relations. Many appreciate the quality of the relationship established with them, with their empathy and humanity, and the patient participants stress this helpfulness.

Now, I have to say this: I have found some marvelous people during the course of my illness . . . Because everyone has done all they could and everyone has tried in any case to help me; they have never given up. And I must say that I have been really lucky . . . (Patient Participant 15)

However in some interviews, a negative dimension arises about the patient–staff relationship which is to do with a “lack of attention,” to haste, to the insufficient resources:

But I must be absolutely straight with you, I said: these people have not even taken the commitment seriously. . . They have just given me some pills that I buy. . . It’s not enough! Here the Doctor is always busy here and busy there, she never stays very long. (Patient Participant 6)

### **The need to receive information**

In the complex and unusual diagnosis and treatment process, patients express their need to receive constant and understandable information. Some are able to express this need to the professional health care providers whereas others find it more difficult to do so.

In the interviews, patient participants stress the professional health care providers’ ability to use language that is understandable and suited to the patient:

Because it is true that patients are often annoying! Petulant, they always ask the same things. But once you have understood that that patient has that characteristic . . . it is necessary to give that patient suitable answers! (Patient Participant 12)

Instead some patients reveal that their need to receive information is a critical issue.

Maybe what’s missing, but I don’t know to what extent, is precisely to give more information about your illness . . . Also to know why. Because, well . . . perhaps they don’t know much more than we do. (Patient Participant 13)

Taken as a whole, the means used so far to provide patients with information are in any case perceived as satisfactory.

## **Results of the Interviews With the Professional Health Care Providers**

### **Dealing With the Disease**

#### **The patient’s experience**

It emerges from the interviews that the professional health care providers are attentive to recognizing the difficulties patients have in dealing with a diagnostic procedure that is often long and tiring. They stress how the patients arrive at the center after an experience marked by multiple tests, doctors, experiences in various hospitals. When our professional health care providers meet these patients, they consider it important to take their history into account, to be delicate and patient, to devote time to them:

There is no doubt that the fact of having a rare disease makes it even harder for people to find their way because in effect it is not easy to arrive at these diagnoses. (Professional Health Care Participant 1)

The professional health care participants recognize in the patients an experience of pain, suffering, linked also to the difficulty of managing everyday life because of their symptoms or invalidating conditions which have a strong impact on their personal choices:

Already just the fact of working or resting. If I still have a job, with the crisis that there is in the job market, these people have to take days off, also several times a week to do their treatment, perhaps they have

every right to do so but sometimes they say: “I went to work even though I did not feel at all well so as not to lose my job.” (Professional Health Care Participant 3)

Rare diseases are a set of conditions that bring patients together but which also at the same time profoundly mark those who have them. The same rare disease may bring together some very different experiences. In their interviews, the professional health care provider participants often refer to the multiple types of participants who come to the Center and to the different means of interpreting the disease as regards the various expectations, experiences, and requirements:

I have two patients P. and A., one is nearly thirty, the other is about thirty, and they have the same disease but they experience it in a different way. One distances herself from the illness, sometimes she comes, sometimes she doesn't . . . the other tries to find other approaches, other drugs, looking also outside Italy. (Professional Health Care Participant 4)

### **The specific nature of rare diseases**

Some interviewees point out how difficult it is to speak of rare disease because it is a general term, based on an epidemiological criterion, which covers very different pathologies, each one with broad spectrums of possible disease expression. In some interviews, there is an attempt to identify some specificities in the experiences of patients with rare diseases which sets them apart from other types of patients:

The problem of rarity is a problem of a certain weight: the problem is that the pharmaceutical companies do not make drugs, i.e. orphan drugs, for them; there is the fact that doctors do not know the disease, even though in reality it is often not like that because many rare diseases are less unknown than what people say, but instead some really are; and there is the fact that they cannot find other patients and so there is the sense of solitude that patients often find themselves experiencing. (Professional Health Care Participant 5)

### **The chronic nature of the disease**

The professional health care participants describe the chronic and degenerative course of these pathologies and their seesaw progress with periods of remission alternating with periods of relapse and so constant checks are required together with long-term treatment. In particular, the professional health care provider participants stress how, as it is impossible to cure these diseases, it is important to keep them constantly under control, to prevent or to intervene swiftly on possible worsening:

You have to be a sort of bloodhound with the patient, I don't know how to explain it. You must never let him go. Maybe he will tell you he is well, or he reports something to you that may not mean anything. But it could be a complication, the beginning of a serious problem. If you overlook it, the prognosis could get worse . . . (Professional Health Care Participant 12)

In these moments, the professional health care participants themselves can feel discouraged, sorry for the patient, and, in some cases, defeated because they have not been able to prevent the worsening of the disease.

### **Treatment management**

The interviews reveal the complexity of the treatment choices and management. Many of those interviewed explain how it can be difficult to swiftly identify the best treatment approach: Often there are few studies to refer to and there is a subjective variation in the response to a drug.

For us it is probably easier to find a treatment for a patient who has a common illness because it can be studied better, rather than to give one to a patient who has a disease that only another ten people in the world have. There are no studies; we are blundering about in the dark . . . (Professional Health Care Participant 11)

Often it may be necessary to vary the treatment in time. Often the choices have to be negotiated with the patient.

### **Dealing With Expectations**

#### **Admission and follow-up**

The professional health care participants interviewed report how it can be difficult to find someone to follow these patients, be it a specialist or an appropriate center. This may be because patients seek someone who can take them on globally, with a holistic approach which is not exclusively linked to individual problems:

Usually patients with a rare disease arrive in our Center after a long period through other centers and with other doctors; they already have an experience of disease, tests, suffering. They arrive here with a lot of expectations. (Professional Health Care Participant 6)

#### **Being of assistance**

According to the professional health care participants, patients have great expectations from the professional health care providers of the rare diseases Center: They hope to find a stable reference for their problems, they expect a clear and conclusive diagnosis, they hope to receive suitable treatment and to be followed in the progression of their disease:

They want a lot of confirmation from the doctors about the treatment, the tests. But I find that they are followed in an exceptional way. There is a lot of attention to the patient, he is not lost. Even a patient who does not show up for a while is called to find out how he is doing. There is a lot of attention, I repeat, and I have never seen that in other places. (Professional Health Care Participant 3)

#### **Inadequacy and frustration**

The interviews reveal how difficult it is to live up to the patient participants' expectations. The diagnosis is not always reached in a short time or it may not be possible to reach one at all, and the treatment is not always effective or does not guarantee the result desired by the patient:

Above all they want a diagnosis. Sometimes we are able to find our way in the field of pathology, understand for example that this pathology is autoimmune, that it is necessary to prescribe an immunosuppressive treatment . . . for a certain period we say that it is necessary to 'freeze' the diagnosis in a certain sense, not to give it a name immediately. (Professional Health Care Participant 10)

The professional health care provider participants consider the subject of recovery to be particularly delicate. They know that some patients do have this hope and that it is highly unlikely to happen as these are chronic pathologies.

Being unable to meet patients' expectations can sometimes entail an experience of frustration and disappointment both in the patient and in the professional health care provider.

We aren't always able to give a diagnosis. . . . These patients remain a bit not so much discouraged as disappointed. They come to us and they expect that there is a cure for this disease and instead there isn't

one, because it has not yet been discovered or because it does not exist! (Professional Health Care Participant 7)

Sometimes the expectations of these patients turn into expectations toward the professional health care staff and the Public Health System, from which a great deal is expected in terms of attention and services:

Some live with the impression that the rare disease is something different from other diseases, when this is not the case. Some think: "If I have this disease everyone owes me: I must look after myself, you must look after me and you must give me these drugs free." (Professional Health Care Participant 12)

### Building Relationships

#### Features of the relationship

The patients who come to the Center are followed for very long periods. They can even have several appointments per month for treatment and check-ups. This continuity of relationship facilitates lasting relations. Some professional health care participants describe the relationships that can become established between themselves and the patients very positively thanks to the trust and sometimes friendship that is created. They point out how this aspect can have a positive influence on the management of the patient himself:

I really see that sometimes for the patients, above all those with rare diseases, it is necessary to have some words of comfort . . . If this human aspect is missing I notice that the patients have more difficulty, even in understanding the treatment, and understanding, for example, why there is no drug available. (Professional Health Care Participant 9)

Some stress how there is a certain variability in the relations, depending on the character of the person or of the professional health care provider and in the way both of them deal with and conceive the disease. This can make empathetic interaction and mutual understanding difficult.

Sometimes a situation of mutual dependence is created, with the risk that the doctor cannot always maintain the necessary lucidity to deal with situations and for the patient to feel lost when he is entrusted to another doctor:

Instead often the patient becomes dependent on the doctor. We see this here where in any case we are a university Centre, so the doctors who follow the patients directly, who are the doctors doing their specialization period, change each time. The patient feels lost. (Professional Health Care Participant 5)

Some professional health care participants point out the risk that they may end up by being too involved, for example, by allowing the patient to contact them at home, becoming a point of reference also for problems that are not directly inherent to the rare disease, and they suffer detachment strongly.

. . . Worried because then among other things I do not know whether I made a mistake or not; I gave a number . . . my mobile number, my personal one. So nearly every Saturday or Sunday I was called . . . That anyway always . . . always in touch, for whatever reason! (Professional Health Care Provider Participant 12)

### Relationship with informed patients

Many professional health care participants stress the difficulties encountered with working with patients who are often very well informed about their pathology. They explain how these patients gather a vast

amount of information on the Internet, but that they do not always have the competence to analyze this information in a critical way. This situation can give rise to some problems because of the heterogeneity of the sources and because of an approach to the disease which is at times different from that of evidence-based Western medicine. It can become difficult to establish relationships of trust with informed patients because the medical staff are assessed on what they say about the disease and its treatment. Often professional health care participants explain how important it is to be perceived as honest and sincere, and to propose to the patients that they should discuss together the information gathered:

Sometimes the information becomes misleading, above all the info found without suitable instruments, on internet, "googling" the name of the disease . . . decoding that mass of information that the patient has found on internet and seeing whether he has really understood what is accepted in the scientific community . . . Sometimes I say that he should print it for me and we can talk about it together. (Professional Health Care Participant 8)

### **Family involvement**

The interviews reveal how the professional health care provider participants recognize the important role that the family plays for their patients, providing a fundamental support both for the emotional aspects and in the management of everyday living.

Family members often accompany the patient for the check-up or treatment sessions and ask to sit with them, to be informed on how the treatment and the disease are progressing. The professional health care provider participants consider it important to involve them while always respecting the limits of privacy.

The family is considered a good resource: They know the disease of their family member very well and often they are the first to notice any changes, signals of possible worsening, and to tell the doctor:

The patient's family notice changes. Take a patient who is well and who then at a certain point does not want to go out any more, and sleeps all the time, then maybe the wife will call me to tell me that there's something wrong. I think the patient's family are little experts on the disease . . . (Professional Health Care Participant 6)

However, some professional health care provider participants point out how sometimes it is difficult to relate with the family because they display a level of anxiety and worry about the condition of the patient's disease that is often worse than that of the patient himself:

Often the family asks more questions than the patient himself, perhaps the patient is a bit disorientated, only attentive to his symptoms . . . he talks about what he experiences. Instead the family wants to know all about the impact this can have! In other words, whether this disease can be hereditary; above all if they have any children, if it can be transmitted, whether anyone in the family might already have it or not . . . they want to be reassured too. (Professional Health Care Participant 11)

### **Being Operators in the Context**

#### **Relations with other professional health care providers**

The interviews reveal how the CMID is a Center where various professionals are at the disposal of the patient to take care of him. In general, the professional health care provider participants report a serene work environment and good relations among all the professional staff. They point out how mutual willingness to help, cooperation, and the opportunity to compare notes are positive aspects:



If there is a person who can't give you an answer on the spot you can soon have a chance to check with them. Colleagues are very helpful, also staff at different professional levels, willing to talk to each other, to tell each other about issues concerning the patients. It's positive, you don't feel alone or at the mercy of situations that you don't know how to deal with in that moment. (Professional Health Care Participant 1)

In particular, what emerges as being important for the medical team is the multidisciplinary aspect. The presence of doctors at the Center with different specializations makes it possible to tackle the various multi-organ problems that rare and chronic diseases can entail. The professional health care participants stress the usefulness of being able to discuss cases together so as to better investigate the situation of a patient and they explain how this is a service also for the patients who can thus consult with specialists directly without having to go to another Center:

As this is a multidisciplinary Centre, with many specialists, we can observe the patient from various viewpoints: this enables us to understand the patient and his disease better. For the patient I think it is a positive aspect too because it means he is not passed around like a parcel between various doctors, he trusts us and so he entrusts himself to us . . . (Professional Health Care Participant 1)

On the other hand, some professional health care provider participants stress how integrating the various specialists is not so easy: One may be missing who would be the point of connection between the various points of view.

The nursing staff instead express the need for greater involvement within the medical team as regards the diagnostic, treatment, and support process:

Being involved . . . today we talk about this patient, what his prospects are, programmes, not only from the point of view of the treatment but also the general picture of the patient. I know this has been done, between the doctors and ourselves we have been a bit left out. This is not a criticism, it is just that I would like to be involved a little bit more in the general picture of the patients. (Professional Health Care Participant 3)

### **Managing difficulties**

The interviews reveal that there are some aspects that the professional health care provider participants consider to be critical in the organization of the CMID. They feel the need to have greater spaces for receiving patients, both to guarantee greater privacy and intimacy during interviews, and so as to better manage the moments devoted to infusion therapy in the day hospital:

In a room that is half the size of this one, we have from fifteen to seventeen patients do treat in the eight hours of the day, concentrated all together, and at times you cannot follow them all well. (Professional Health Care Participant 4)

Some professional health care provider participants express regret about the long waiting times that patients have to undergo. They think that these can be disturbing for patients who already have a history of fatigue and anxiety about their uncertain condition. Others fear that an increase in the number of patients can make the difficulties acute. They consider that it would be useful to better define the indications and appropriateness for access to the Center and admission of the patients followed there.

## **The nature of the role**

In their interviews, the professional health care provider participants refer to the various activities that are carried out at the CMID and which can give rise to a certain degree of confusion not only in the patients but also in the external professional health care providers who use it such as the family doctors. At the CMID, three different activities are carried out: bureaucratic work, research, and health care. As regards the bureaucratic side, the Center does administrative work which regards aspects of exemption from payment for treatment and the management of the Register of Rare Diseases. This bureaucratic function means that there are often conflictual aspects between professional health care providers and users who pursue different objectives or who view the Center in different terms, expecting different types of health care:

I have had to do with another aspect of the CMID, the aspect of . . . Center of Coordination of Rare Diseases of the Piedmont Region. This means an activity which is bureaucratic from a certain point of view because it involves inserting these patients in the Regional Register; issuing exemptions and after that a treatment plan if there is one; this means exemption from the ticket (according to the Italian National Health system, those without a ticket exemption have to co-pay for their treatment up to a certain minimum sum—called “ticket”—after which treatment is dispensed free of further charge) for the tests that need to be done and the treatment plan with free drugs . . . And so for many this is something . . . important; for some sometimes perhaps it is more important than the concept of undergoing treatment. (Professional Health Care Participant 12)

As regards the research aspect, nearly all the professional health care provider participants consider this to be at a very high level. The Center is considered a dynamic and innovative study context, an environment able to provide great opportunities and to provide enriching skills, providing opportunities to broaden knowledge.

The moment the issue moves from the research dimension to that of health care, the professional health care provider participants reveal a sort of ambivalence, in particular as regards experimental treatments, between the positive opportunities for research and the critical aspects regarding the administration of experimental treatments, the unpredictability of the risks and consequences; they are well aware that improvement is not guaranteed, nor is it sometimes even possible to assess.

The risk is the unsatisfactory efficacy of a drug which produces a sense of frustration in the professional health care provider participants, particularly when they are faced with deterioration in the patients:

As ours is a centre of documentation on rare diseases, they come to you hoping that you can give them the most suitable treatment that can help them to get better. Instead you have to point out that unfortunately, as these are rare diseases, firstly there are not always codified treatments, and secondly that unfortunately the disease, though it can be kept under control, will not get better. (Professional Health Care Participant 6)

## **Discussion**

People with rare diseases report an experience of suffering, anxiety, and fatigue linked to the phase of onset of the disease. This is caused both by the symptoms, which can be severe, and by the difficult and often long process of finding a diagnosis with many tests, specialist consultations, admission to various hospital wards and institutions. Often patients experience a sense of isolation and disorientation. In literature, it emerges that patients with rare diseases experience delay in identifying a diagnosis as being one of the greatest problems (Faurisson & Kole, 2009). Providing a diagnosis, a doctor might give meaning to the personal sensations of a patients, legitimizing their illness experience (Corbin, 2003).

After diagnosis, and finding a Center where they can be followed, the attention of the patients participants shifts to how to manage the disease on an everyday basis. The condition of their disease being rare is not described or referred to as the most problematic element. A French study has pointed out how these patients do not perceive the aspect of rarity as the greatest source of difficulty, so long as they have found health professionals who are able to recognize their requirements (Huyard, 2009).

The patient participants focus their attention above all on the difficulty of living with symptoms that impact on their daily lives, which can cause limitations or changes in their habits. They also stress the problem of their need for long-term therapy, some of which has serious side effects, and the need to frequently go for check-ups and tests which takes time away from their personal lives.

The problems that emerged, rather than indicating the specific nature of rare diseases, are similar to those of many people who have chronic and degenerative diseases with whom those interviewed share also the problems of a social nature, such as the difficulty of holding down a job, the need to move near the health care Center, the choice made by some family members to devote themselves entirely to looking after the patient, abandoning their job (Faurisson & Kole, 2009).

Living with a chronic, degenerative disease requires a complex process of adaptation to a changeable condition that upsets the order with which a person's existence and perception of self was organized (Kralik, 2002). Many of the patient participants interviewed talk about how they have developed adapting strategies to deal with their new condition of life. They report that these are effective for living with their disease. Despite the presence of chronic and often degenerative diseases, the objective of maintaining a life that is as normal as possible is defended as a value and it is experienced as possible by patients (Joachim & Acorn, 2003). It is the everyday experience, made up of trial and error, that makes patients able to manage themselves in their disease (Kralik, Koch, Price, & Howard, 2004).

Patients conduct a process of re-elaboration to bring the presence of the disease in their lives from an extraordinary event to an ordinary event that they can live with (Kralik, 2002). This is a continuous and not linear process with phases of adaptation and phases of re-negotiating the patient's condition, also as a result of the progression of illnesses that often alternate phases of clinical stability with phases of worsening.

Within the patient's process of the reconstruction of his/her identity and adaptation to the illness, the encounter with the health service world plays a decisive role. According to what the patients have reported, their arrival at the CMID meant finding a stable point of reference in the course of their disease. The patient participants showed that they did not only seek a place where they could receive care and treatment but also a place where they could be welcomed, understood, and involved. For them, it is necessary to build a treatment alliance with the health care staff to deal with the process of adaptation to a life that is altered by the arrival of the disease. What is said and exchanged during the meetings between the patients and the health care staff can influence in a positive or negative way the patient participant's efforts to reconstruct his/her self-image (Telford, Kralik, & Koch, 2006).

The professional health care participants have shown themselves to be particularly sensitive toward the patients' experiences of discomfort and their difficulties. They recognize the suffering caused by the impact of the disease and stress the need to treat patients with helpfulness and delicacy when they come to the Center for Rare Diseases. The staff explain how relationships can develop which are intense, lasting, sometimes relationships of friendship. Following their patients for a long time, they feel they participate in their histories, at times becoming points of reference for the patients even for problems which are not

strictly to do with the disease. The risk is a strong emotional and workload for the health care staff and an excessive involvement which can at times make it hard to maintain lucidity in treatment choices or can make detachment from these patients painful when this occurs.

Often professional health care participants show that they do not have a uniform approach toward patients. In some moments, they seem to place the person at the center of their professional actions, letting an empathetic attitude of attention to the experience of the patient prevail. At others, instead what seems to be paramount is attention to the disease and its treatment, where the specialist contribution is what counts, with the human component in second place. In any case, all the staff stress how building a relationship of trust with the patients can be a strong point in the course of treatment, guaranteeing better compliance by the patient with the treatment proposed.

According to the professional health care participants, the admission of a patient who comes to the CMID is a delicate moment. They feel very strongly the burden of the patient's expectations in terms of receiving a diagnosis, effective treatment in a short time, and prospects for the future. Patients often have absolute faith in medical science's ability to solve any kind of problem, and when this does not happen, as one Italian study has suggested, this easily unleashes reactions of a lack of confidence in a health system that is considered not to be up to the job (Cipolletta & Oprandi, 2014). Moreover, the impossibility of responding to such high expectations generates a sense of frustration both in the health care staff and in the patients.

One complex aspect that has emerged in the interaction between health care participants and patient participants, which is fundamental in building an effective treatment strategy, is that patients are increasingly well informed. Patient participants involved in the study report that they seek information that can help them to understand the progression of the disease and the treatment possibilities. Sometimes this quest is carried out in a way that is independent from the health care staff, via Internet, reading texts, and going to conferences on the subject. The information gathered is then measured against what the health care staff provides.

The possibility of developing knowledge of their disease helps patients to continue to exercise direct control over their lives and over treatment. They can exercise a greater power in the interaction with the medical staff (Åsbring & Närvänen, 2004). However, the information patients find independently is not always understood, and if it is not accompanied by explanations, it can be disorientating. In literature, it is reported how often patients would like a guide to interpret the information they collect, expecting to receive explanations that they can understand (Huyard, 2009)

The professional health care participants show as patient can have access to a mass of information which he is not able to select and it can lead to creating false expectations. They report how their approach is to propose that they can act as mediators, inviting the patient to discuss with them what they have found so that they can understand it better. If, at times, discussion with informed patients can test the doctor's decision-making autonomy, today this is already recognized as an element that facilitates communication and the building of a relationship of collaboration with the health care staff (Aymé, Kole, & Groft, 2008).

One aspect that has been identified as being complex regards the management of treatment for these diseases. The doctors explain how it can be difficult to swiftly identify the best treatment approach, as it is not evident in literature or because of the variability in individual response. This is not always understood by patients who want to obtain improvements rapidly. Moreover, doctors also report how—in a period of clinical stability—there may be negotiation with the patient about the quantity of drugs to take. Having to deal with patients who are often experts on their disease, or in any case people who know very well their

individual response to drugs taken over a long period, at times leads the medical staff to discuss their treatment choices, and produce clear arguments or to give in to some requests. Doctors find that they have to take decisions together: with the patient they achieve a dynamic process, in which both parties make an effort to understand the needs and wishes of the other, in the context of a relationship of trust and respect (Lown, Clark, & Hanson, 2009).

We realized the study in a Regional Center of Reference for Rare Diseases part of a clinical and epidemiological network. According to our patients participants, their arrival at this Center meant finding a stable point of reference in the course of their disease. For some this coincided with getting a diagnosis, for others it was a treatment process, for many this had the added advantage of ease of access to medical staff, check-ups, tests, and treatment in a single place, thus saving time and energy. For most patients the positive experiences regarded being able to establish a treatment relationship with the health care staff who are considered reliable and qualified. We suggest that patients want to find a Center that will admit them and follow them up in a holistic way, managing all aspects of their treatment. According to a German study, the associations of patients with rare diseases consider that the care of these patients should be based precisely on a holistic, multidisciplinary approach centered on the patient (Reimann, Bend, & Dembski, 2007). Increasingly, patients with chronic disease express the need to feel welcomed, understood, and involved in the treatment as they have to have a constant relationship with the various services. The perception of the quality of the care received often depends on the degree of involvement the patients have experienced in the system (Staniszewska & Henderson, 2005).

## **Conclusion**

We wanted to explore the illness histories of patients and the experiences of the professionals who follow them, compare their portrayals and evaluate the strong and weak points of the course of treatment, creating the conditions for introducing elements of change that are considered useful for improving the course of treatment.

From the results of the study it may be seen how, according to the experiences of the professional health care participants, the problem of disease rarity coincides with the difficulties expressed by the patients in reaching a diagnosis and in finding someone to follow them. Every day the professional staff find themselves up against the suffering caused by the impact of the disease on the lives of the patients and this also involves them emotionally, giving rise to feelings of impotence, frustration, and discouragement. The professional health care participants recognize as a further element of complexity in the relationship the fact that many patients are expert and aware of their own diseases and that this often imposes that the treatment choices have to be revised and more in general the means of interaction with the patient have to be orientated toward discussing the decisions of the clinical course of the health care. Every professional who works in this environment has to concentrate all the time on understanding also the patient's unexpressed needs. Attention to the reaction of each individual patient, rather than proposing standardized solutions that cannot always suit everyone, can make it possible to fully understand the needs of each patient and to provide personalized strategies to satisfy those needs. The professional health care participants point out that a strong point of their work is the multidisciplinary approach that is adopted, which involves different specialists in a climate of cooperation and helpfulness in seeking the best treatment options possible (Kodra et al., 2007).

The patient participants had to deal with numerous difficulties in their disease history. They experience as a significant moment the time when they found a point of reference for their treatment: A single place where

they could be followed for medical advice, treatment, tests and check-ups, where they could feel welcomed and recognized.

The subject of the rarity of a disease, once there has been a diagnosis and a treatment center has been identified, is not considered the most problematic element by the patients and by the health professionals.

We conducted the study in a single Center of reference for rare diseases in a region of North Italy. The patients involved, followed as outpatients, were self-sufficient and with autoimmune type pathologies. The experiences and considerations that have emerged can therefore only be partially extended to other contexts. Moreover, the pathologies of the patients interviewed are different, and even though they have some common characteristics (chronicity, potentially invalidating, frequent treatment, and repeated tests), they make the experiences of the participants only partially applicable to other Centers.

We provided the opportunity to promote attention on the subject of rare diseases. To understand the needs and difficulties encountered by people with these pathologies, together with the experiences of the health care staff who take care of them, has made it possible to identify areas that may be useful for improving the quality of the health care services.

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**Table 1.** Characteristic of the Sample.

	Patient Participants	Professional Health Care Provider Participants
Gender	16 women, 6 men	10 women, 2 men
Age (years)	<i>M</i> = 56 (range = 21–79)	<i>M</i> = 38 (range = 27–54)
Follow up CMID (years)	<i>M</i> = 4.63 (range = 1–14)	
Period of service at CMID (years)		<i>M</i> = 3.8 (range = 6 month–13)

Note. CMID, Multidisciplinary Center of Immunopathology and Documentation of Rare Diseases.



**Table 2.** Themes and Essential Elements That Emerged From Patient Participants' Interviews.

Themes	Essential Elements
Dealing with disease development	Getting a diagnosis Treatment approach Seesaw progression between remission and relapse Predicting the future
Living with the disease	Adaptation strategies The quest for independence The need to know the disease
Everyday living	Functional limitations Experiencing pain Influence on work
Relating to others	The role of the family Comparison with other patients
Relations with professional health care providers	Admission and follow-up Features of the relationship The need to receive information

**Table 3.** Themes and Essential Elements That Emerged From Professional Health Care Professional Participants' Interviews.

Themes	Essential Elements
Dealing with the disease	The patient's experience Specific nature of rare diseases The chronic nature of the disease Treatment management
Dealing with expectations	Admission and follow-up Being of assistance Inadequacy and frustration
Building relationships	Features of the relationship Relations with informed patients Family involvement
Being operators in the context	Relations with other professional health care providers Managing difficulties The nature of the role